



DVL3 gene

dishevelled segment polarity protein 3

Normal Function

The *DVL3* gene provides instructions for making a protein that plays a critical role in development before birth. It is one of three DVL genes in humans (*DVL1*, *DVL2*, and *DVL3*). The proteins produced from these genes work together in chemical signaling pathways known as Wnt signaling. These pathways control the activity of certain genes and regulate the interactions between cells during embryonic development. Signaling involving the DVL proteins appears to be important for the normal development of the brain, skeleton, and many other parts of the body.

Health Conditions Related to Genetic Changes

Robinow syndrome

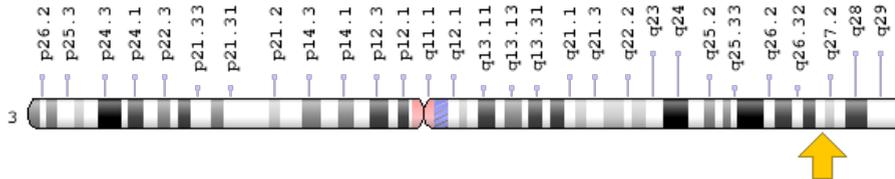
At least six mutations in the *DVL3* gene have been found to cause the autosomal dominant form of Robinow syndrome, a condition that affects the development of many parts of the body, particularly the skeleton. Autosomal dominant inheritance means that one copy of the altered gene in each cell is sufficient to cause the disorder.

All of the identified *DVL3* gene mutations occur near one end of the gene and are predicted to shorten the DVL3 protein. Researchers are working to determine how these changes affect the protein's function. The changes may have a dominant-negative effect, which means that the altered protein produced from one copy of the *DVL3* gene interferes with the function of the normal protein produced from the other copy of the gene. Alternately, the changes may have a gain-of-function effect, giving the altered protein a new, as-yet-undetermined function. Either way, the abnormal DVL3 protein likely impairs Wnt signaling. Problems with Wnt signaling pathways disrupt the development of many organs and tissues before birth, leading to Robinow syndrome.

Chromosomal Location

Cytogenetic Location: 3q27.1, which is the long (q) arm of chromosome 3 at position 27.1

Molecular Location: base pairs 184,155,311 to 184,173,614 on chromosome 3 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- dishevelled 3 (homologous to Drosophila dsh)
- dishevelled, dsh homolog 3
- DRS3
- KIAA0208
- segment polarity protein dishevelled homolog DVL-3

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Database: The Wnt Gene Family and the Evolutionary Conservation of Wnt Expression
<https://www.ncbi.nlm.nih.gov/books/NBK6212/>
- The Wnt Homepage, Stanford University
<https://web.stanford.edu/group/nusselab/cgi-bin/wnt/>

Clinical Information from GeneReviews

- Autosomal Dominant Robinow Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK268648>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28DVL3%5BTIAB%5D%29+OR+%28dishevelled+segment+polarity+protein+3%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- DISHEVELLED 3
<http://omim.org/entry/601368>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_DVL3.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=DVL3%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report#!/hgnc_id/HGNC:3087
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:1857>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/1857>
- UniProt
<https://www.uniprot.org/uniprot/Q92997>

Sources for This Summary

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